XL 4q12 DC

Deletion/Fusion Probe

Order No.:
D-5123-100-OG

Description

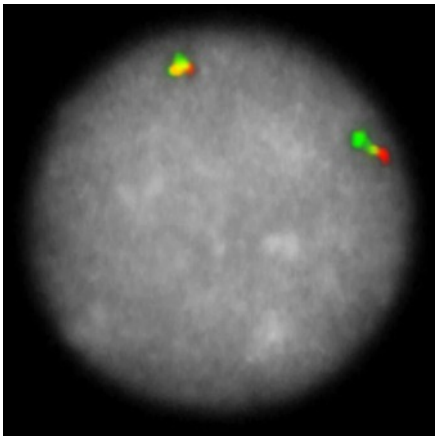
XL 4q12 DC is a dual color probe which detects rearrangements and deletions at 4q12. The probe contains an orange probe hybridizing to the CHIC2 gene region. A green probe hybridizes proximal to FIP1L1, and another green probe hybridizes to PDGFRA and the region distal to the gene. XL 4q12 DC is a two color alternative to XL 4q12 D-5063-100-TC.

Clinical Details

The updated (2016) World Health Organization (WHO) classification of tumors of hematopoietic and lymphoid tissues indicates the category myeloid/lymphoid neoplasms with eosinophilia and rearrangement of PDGFRA, PDGFRB, FGFR1, or with PCM1-JAK2. Eosinophilia is a condition in which the number of eosinophilic granulocytes in peripheral blood or tissue is increased above the normal level. The hypereosinophilic syndrome (HES) is a rare hematologic disorder characterized by a marked and persistent increase of eosinophilic granulocytes in blood or tissue accompanied by organ damage. HES is associated with neoplastic (primary) or reactive (secondary) processes. Patients suffering from hematologic disorders with eosinophilia are typically diagnosed with chronic eosinophilic leukemia (CEL), myeloproliferative neoplasms (MPN), certain variants of acute myeloid leukemia, systemic mastocytosis and others. The most frequent aberration detected in CEL (10-20%) is the interstitial deletion of the CHIC2 gene with breakpoints in the FIP1L1 and PDGFRA genes. The deletion of a fragment of about 800kb is resulting in the FIP1L1-PDGFRA fusion gene, a constitutively activated tyrosine kinase, transforming hematopoietic cells. Other aberrations associated with CEL are translocations generating fusion genes involving PDGFRA. PDGFRA and PDGFRB fusions are sensitive to Imatinib, a proven treatment for BCR-ABL–positive chronic myeloid leukemia, whereas FGFR1 mutations are resistant.

Literature:

- Cools et al (2003) N Engl J Med 348:1201-1214
- Pardanani et al (2003) Blood 102:3093-3096
- Valent et al (2012) Expert Rev Hematol 5:157-176



XL 4q12 hybridized to lymphocytes, one normal interphase is shown. The expected normal signal pattern is two green-orange colocalization/fusion signals, representing two non rearranged FIP1L1-CHIC2-PDGFRA loci. An interstitial deletion of the CHIC2 gene is resulting in one green signal and one green-orange colocalization/fusion signal. PDGFRA translocations are indicated by two green-orange colocalization/fusion signal plus one seperated green signal.

Clinical Applications:

- CML/MPN

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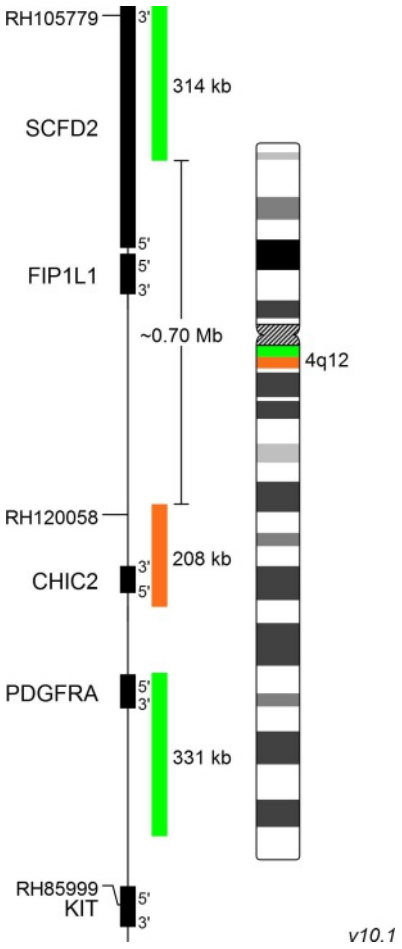
Probes



Related Products

Product	Size	Order No.
XL 4q12	100µl	D-5063-100-TC

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